

### Abstract

This invention is related to novel non-nucleic acid probes, probe sets, methods and kits pertaining to the detection, identification or quantitation of human chromosomes X, Y, 1, 2, 3, 4, 6, 7, 8, 9, 10, 11, 12, 13, 16, 17, 18, 20 and/or 21. The non-nucleic acid probes, probe sets, methods and kits of this invention are particularly well suited for use in multiplex ISH and FISH assays wherein each of chromosomes X, Y, 1, 2, 3, 4, 6, 7, 8, 9, 10, 11, 12, 16, 17, 18 and/or 20, as well as 13/21 as a pair, in a sample or cell can be individually detected, identified or quantitated in the same assay. Multiplex ISH and FISH assays are possible because two or more of the probes used in the assay are labeled with one or more independently detectable labels. Preferably, the independently detectable labels are independently detectable fluorophores. In preferred embodiments, one or more of the probes comprise two or more linked independently detectable moieties wherein the combination of the two or more independently detectable moieties is used to detect, identify or quantitate a particular probe/target sequence hybrid. The methods, kits, probes and probe sets of this invention are particularly well suited for automated analysis, including a slide scanner based system, microscope and CCD camera or a flow cytometer. Furthermore, this invention is particularly useful for detection and identifying chromosome abnormalities such as aneuploidy and polyploidy karyotypes and particularly for preimplantation diagnosis, for prenatal screening and for clinical diagnostic applications.

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